

CLAIMS:

1. An isolated nucleic acid encoding UDP-N-acetylglucosamine: galactose- β 1,3-N-acetylgalactosamine- α -R / N-acetylglucosamine- β 1,3-N-acetylgalactosamine- α -R β 1,6-N-acetylglucosaminyltransferase (C2/4GnT) or a fragment hereof.

2. An isolated nucleic acid as defined in claim 1, wherein said nucleic acid is DNA.

3. An isolated nucleic acid as defined in claim 2, wherein said DNA is cDNA.

4. An isolated nucleic acid as defined in claim 2, wherein said DNA is genomic DNA.

5. An isolated nucleic acid as defined in claim 1, wherein said nucleic acid comprises the nucleotide sequence of nucleotides 1-2319 in SEQ ID NO:1 or sequence-conservative or function-conservative variants thereof.

6. An isolated nucleotide sequence comprising nucleotides selected from the group consisting of nucleotides 1-245; nucleotides 246-435; and nucleotides 436-2319 of SEQ ID NO:1 that hybridizes to a nucleic acid under stringent conditions.

7. A nucleic acid which hybridizes under conditions of high stringency with the nucleic acid having the sequence of nucleotides 1-2319 in SEQ ID NO:1.

8. A nucleic acid vector comprising a nucleic acid sequence encoding C2/4GnT or fragments thereof.

9. A vector as defined in claim 8, wherein said sequence comprises the nucleotide sequence of nucleotides 1-2319 in SEQ ID NO:1 or sequence-conservative or function-conservative variants thereof.

10. A vector as defined in claim 9, wherein said sequence encoding C2/4GnT is operably linked to a transcriptional regulatory element.

11. A cell comprising a vector as defined in claim 8.

12. A cell comprising a vector as defined in claim 10.

13. A cell as defined in claim 12, wherein said cell is stably transfected with said vector.

14. A cell as defined in claim 11, wherein said cell produces enzymatically active C2/4GnT.

15. A cell as defined in claim 11, wherein said cell is selected from the group consisting of bacterial, yeast, insect, avian, and mammalian cells.

5 16. A cell as defined in claim 14, wherein said cell is selected from the group consisting of bacterial, yeast, insect, avian, and mammalian cells.

17. A cell as defined in claim 16, wherein said cell is Sf9.

18. A cell as defined in claim 16, wherein said cell is CHO.

19. A method for producing C2/4GnT polypeptides, which comprises:

(i) introducing into a host cell an isolated DNA molecule encoding a human C2/4GnT, or a DNA construct comprising a DNA sequence encoding C2/4GnT;

(ii) growing the host cell under conditions suitable for human C2/4GnT expression; and

(iii) isolating C2/4GnT produced by the host cell.

15 20. A method as defined in claim 19, wherein said enzymatically active C2/4GnT is selected from the group consisting of:

(i) a polypeptide having the sequence of SEQ ID NO:2;

(ii) a polypeptide consisting of amino acids 31-438 of the sequence of SEQ ID NO:2;

20 (iii) a fusion polypeptide comprising at least amino acids 31-438 of the sequence of SEQ ID NO:2 fused in frame to a second sequence, wherein said second sequence comprises an affinity ligand or a reactive group; and

(iv) function-conservative variants of any of the foregoing.

21. A method for the identification of DNA sequence variations in the β C2/4GnT gene, comprising the steps of:

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- (i) isolating DNA from a patient;
 - (ii) amplifying C2/4GnT genomic regions by PCR; and
 - (iii) detecting the presence of DNA sequence variation by DNA sequencing, single-strand conformational polymorphism (SSCP) or mismatch mutation.
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